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Letter to the Editor

Medulloblastoma in Patient With Rubinstein-Taybi Syndrome

To the Editor:

The following is a brief description of a patient with Rubinstein-Taybi syndrome (RTS) and medulloblastoma. We report the case in recognition of the apparent excess number of brain tumors in patients with RTS [Miller and Rubinstein, 1995] and because of the delay in diagnosis that occurred in this patient.

The patient is a 9-year-old boy diagnosed at age 2 years with RTS on the basis of downward slanted palpebral fissures, micrognathia, broad toes and thumbs, mental retardation, and a stiff, unsteady gait. He was diagnosed with a posterior fossa medulloblastoma in March of 1990. Before the diagnosis, multiple visits were made to his healthcare providers to evaluate recurrent complaints of neck pain, vomiting, and constipation. Those visits focused on evaluations of the neck and gastrointestinal tract. At the time of diagnosis, the tumor had metastasized to the suprasellar region and the left subfrontal area.[1]

DISCUSSION

The literature describing RTS indicates 76% of these patients have a "stiff, unsteady gait" [Jones, 1988]. The patient's ataxia and gait abnormalities were initially thought to be a component manifestation of the RTS. This led to a delay in the evaluation for a possible intracranial mass, postponing the diagnosis of the posterior fossa tumor.

In his review, Dr. Miller noted that 5% of the known cases of RTS have developed benign or malignant tumors, several of those being intracranial [Miller and Rubinstein, 1995]. The interaction of symptoms from a congenital syndrome such as RTS with a second underlying illness such as an intracranial tumor make early diagnosis of the tumor difficult. The increased incidence of tumors in these children suggests that unusual, persistent complaints such as, "my child is not walking like usual," or subtle abnormal physical exam findings should motivate early, thorough evaluation to rule out benign or malignant tumors.

REFERENCES

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Miller RW, Rubinstein JH (1995): Tumors in Rubinstein-Taybi syndrome. Am Med Genet 56:112–115.

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[1] This individual is patient number 9 in Table I of Dr. Miller's study [Miller and Rubinstein, 1995]. This was confirmed in a personal telephone conversation with Dr. Rubinstein.